

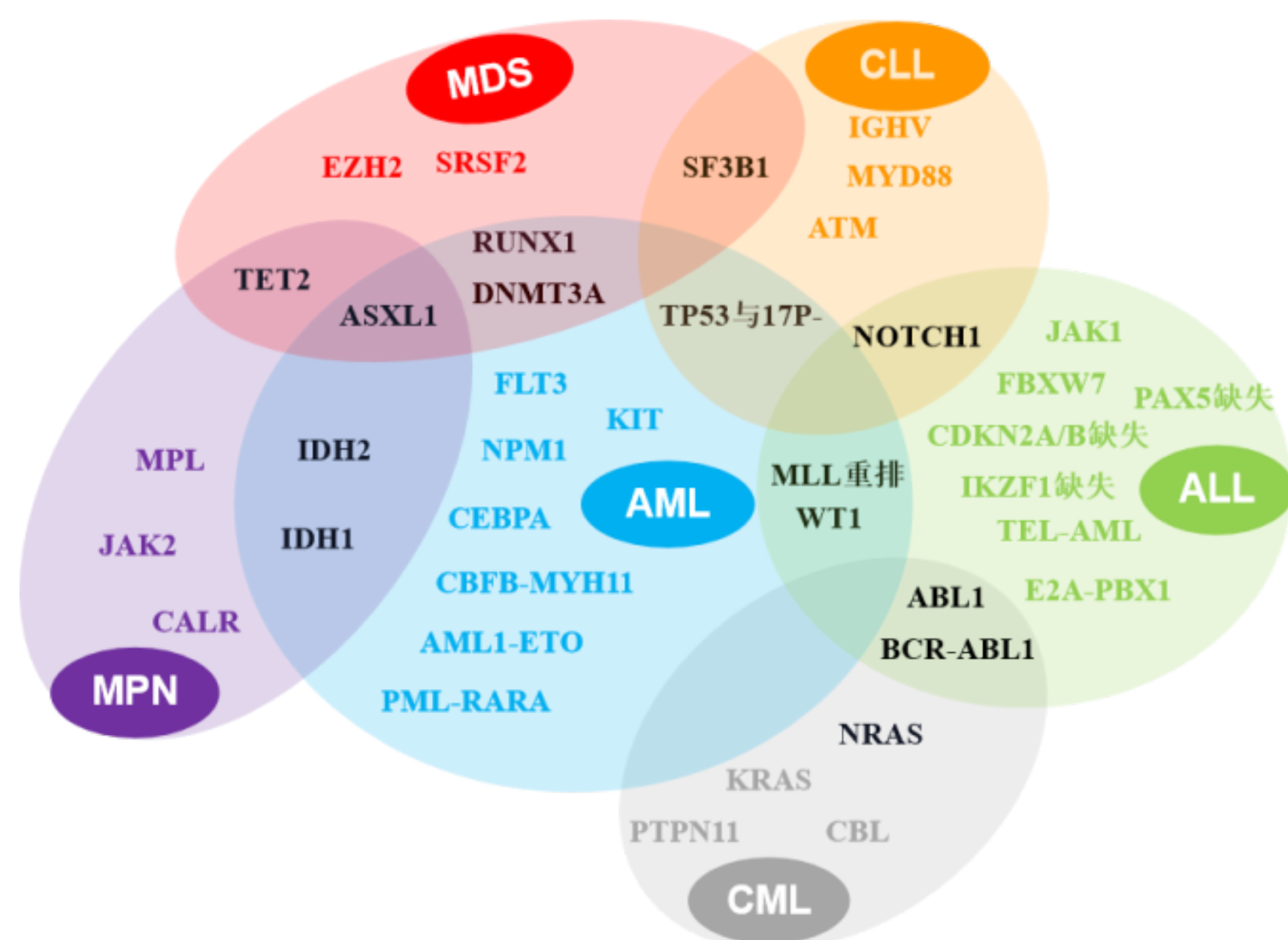
Panel-Ref® Hematologic Malignancies Cocktail Reference Standard--CBP90039

BACKGROUND

Hematological tumors refer to malignant tumors that occur in the blood system (bone marrow, hematopoietic tissue, and lymphoid tissue). Common hematological tumors mainly include various types of leukemia, multiple myeloma, myeloproliferative neoplasms and malignant lymphoma. The overall incidence of multiple myeloma accounts for 10% of hematological malignancies.

INTRODUCTION

CB-Gene has launched a major hematological malignant tumor standard product, involving common gene mutation sites related to MDS/MPN/AML/CML/CLL/ALL and other diseases.



This is a multi-gene and multi-site panel standard, The raw materials used in the mixed samples were basically derived from blood system-related tumor cells. **CB-Gene** has developed a digital PCR (dPCR) detection system for all 38 mutation sites related to 27 genes to accurately determine the corresponding mutation frequency or copy number variation value.

Panel-Ref® Hematologic Malignancies Cocktail Reference Standard		
ABL1	c-MYC	KMT2D
ARID1A	CREBBP	KRAS
ASXL1	EP300	MEF2B
B2M	FLT3	MYD88
BCORL1	IDH1	NF1
BRAF	IDH2	NRAS
BTK	JAK2	PDGFRA
CBL	KIT	PIK3CA
CD79a	KMT2A	TP53

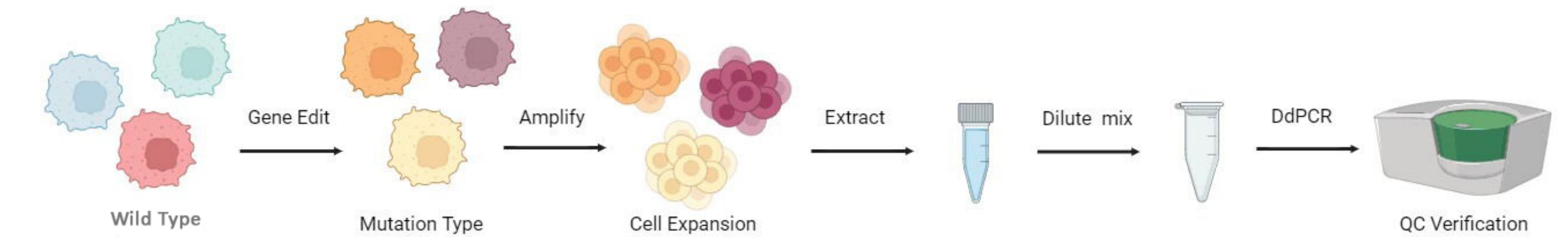
Table 1. 27 genes related to hematological malignancies panel

PRODUCT DATA

This product provides digital PCR results of 38 hotspot mutations (see Table 2 for specific mutation site information)

Gene	Mut.	CDS change	%AF
ABL1	p.T315I	c.944C>T	4.94
ABL1	BCR(E13)-ABL1(E2)	N/A	4.56
MYD88	p.L265P	c.794T>C	4.81
BTK	p.C481S	c.1442G>C	5.25
FLT3	FLT3-ITD(E598_Y599insFD FREYE)	c.1795-1796 ins21(TTGATTCA GAGAATATGAAT)	5.5
FLT3	p.D835V	c.2504A>T	5.92
IDH2	p.R172K	c.515G>A	5.08
JAK2	p.V617F	c.1849G>T	4.68
KIT	p.D816V	c.2447A>T	4.92
KRAS	p.G12D	c.35G>A	5.64
PDGFRA	FIP1L1(E12)-PDGFR A(E12)	N/A	5.63
PDGFRA	p.D842V	c.2525A>T	5.24
PIK3CA	p.H1047R	c.3140A>G	5.96
C-MYC	Amplification	N/A	4.84 copies
CBL	p.Q409fs	c.1227_1227+13del(AGGTACGGATCTAA)	2.79
IDH1	p.R132C	c.394C>T	14.94
NRAS	p.Q61K	c.181C>A	16.7
BCORL1	p.P1681Qfs*20	c.5042delC	10.75
TP53	p.M133K	c.398T>A	0.76
BRAF	p.V600E	c.1799T>A	13.17
ARID1A	p.D1850Tfs*33	c.5548delG	7.65
ARID1A	p.P1115Qfs*46	c.3344delC	7.31
EP300	p.K292Rfs*25	c.875delA	6.95
EP300	p.M1470Cfs*26	c.4408delA	7.34
CREBBP	N/A	c.1824-2A>G	7.75
B2M	p.R117*	c.349C>T	7.24
NF1	p.Y628Tfs*3	c.1882delT	7.63
NF1	p.N2341Tfs*5	c.7022delA	7.47
TP53	p.S215G	c.643A>G	11.8
MEF2B	p.V316Sfs*6	c.944dupG	7.54
CD79a	p.L191_G208del	c.568-2_610del(TCCCAGGGCCTGAACCTG GACGACTGCTCCATGTATGAGGACATC)	3.02
KMT2D	p.G3465*	c.10393G>T	7.24
KMT2D	p.P2550Lfs*33	c.7649delG	7.24
KMT2A	KMT2A-MLLT3(E10-E6,E9-E6)	N/A	2.69
KMT2A	p.D877Pfs*8	c.2629_2630delAG	6.8
KMT2A	p.P773Rfs*8	c.2318delC	6.86
ASXL1	p.G645Vfs*58	c.1926delC	6.98
KMT2A	KMT2A-PTD(E2-E8 Dup)	c.22807bp-Duplication	2.6

PREPARATION PROCESS

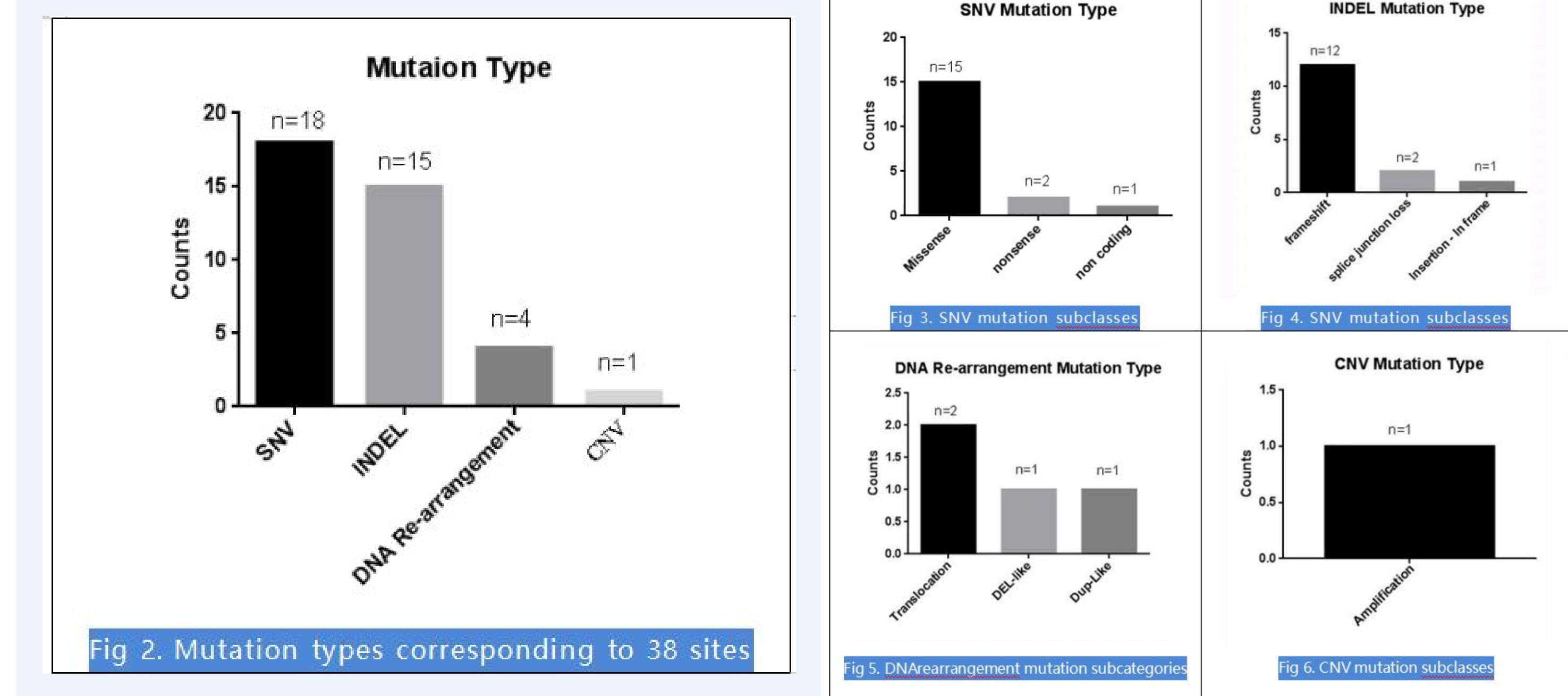


Panel-Ref® Hematologic Malignancies Cocktail Reference Standard preparation process

TEST PRINCIPLE

This product can be tested using high-throughput sequencing (Next generation sequencing, NGS) or digital PCR. The mutation sites and mutations contained in the product can be determined through sequencing data analysis or digital PCR detection signals.

MUTATION TYPE



This panel standard contains many mutation sites related to hematological malignancies, of which 38 sites have been verified by ddPCR, including 18 SNVs (which are divided into missense, nonsense, etc.), 15 INDELs (large Most were insertions and deletions of repeated bases), 4 DNA rearrangements, and 1 copy number variation.